

- Submission of Pharmacogenomic Data to FDA and standards

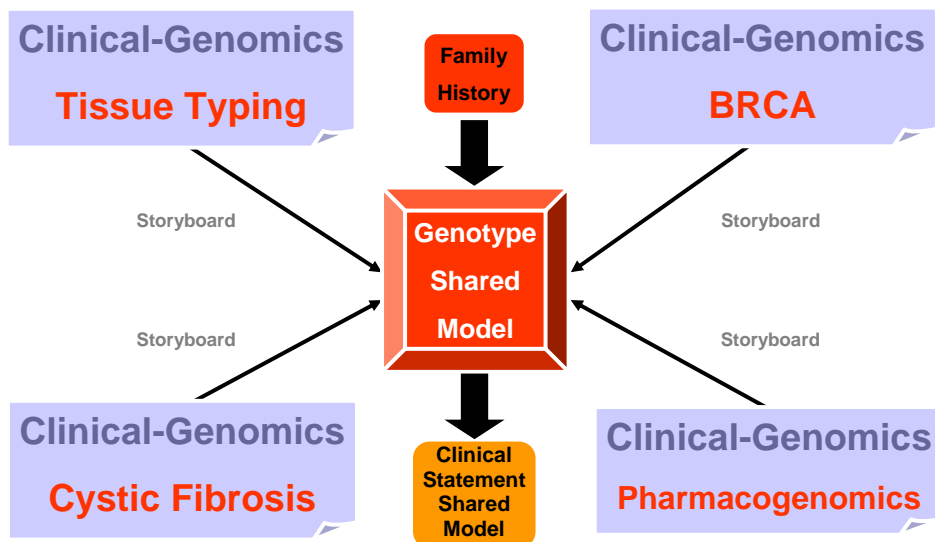
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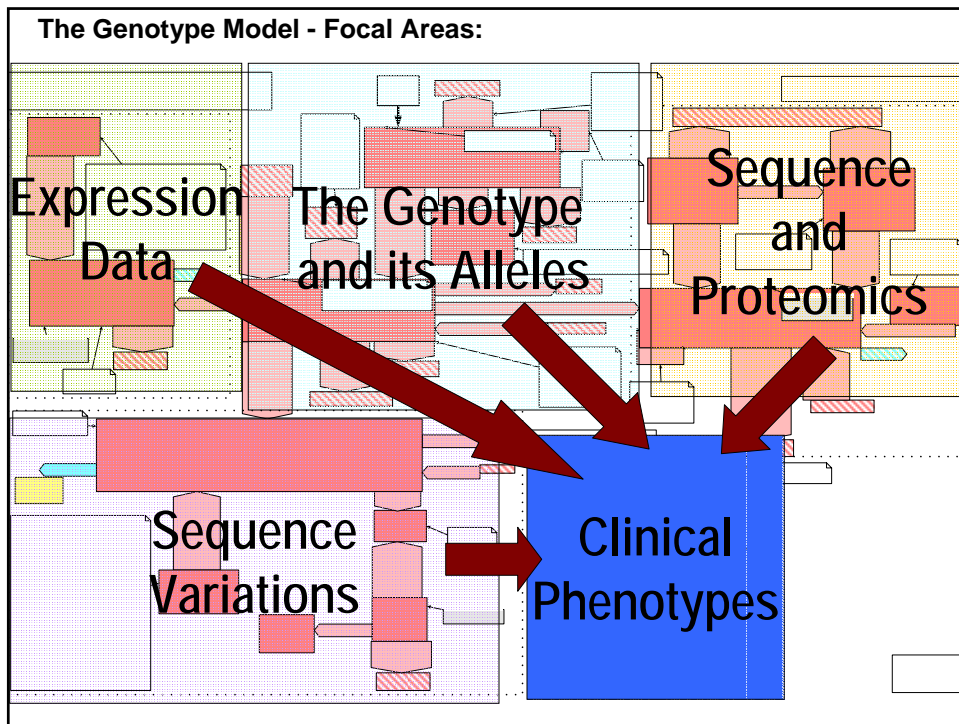
[illegible]

HL7 Clinical Genomics Special Interest Group

- **Mission:** to enable the standard use of patient genomic data such as DNA sequence variations and gene expression levels, for healthcare purposes ('personalized medicine') as well as clinical trials & research.
- CG Specs are in a **DSTU** (Draft Standard for Trial Use) Period; thus, substantive **changes** could take place until it goes to normative edition!!
- Joint work with HL7 RCRIM (clinical trials) on Pharmacogenomics is work in progress

HL7 Clinical-Genomics Current Work





HL7 Clinical Genomics SIG

Document: Genotype Shared Model

Subject: Genomic Data Rev: POCG_DM000020-c Date: J

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IBM Healthcare and Life Sciences

Submission of Pharmacogenomic Data to FDA

- What data should be submitted?
 - o raw data (*cel file, probe set file, image data*)
 - o normalization algorithm
 - o list of genes
 - o biological interpretation of the data
 - o MIAME guidelines
 - o phenotypic information

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ExpressionProperty

expressionProperty

NORMALIZED_INTENSITY, QUALITATIVE_EXPRESSION, ABSENCE

is populated based on the selected code from the vocabulary and its type is then selected accordingly.

For example, if code = NORMALIZED_INTENSITY, then value is a numeric value representing the normalized intensity. If the code = QUALITATIVE_EXPRESSION, then value is a string representing the expression level, and holds either PRESENT or ABSENCE.

description of the allowed vocabularies for codes and their corresponding values could be found in the specification.

EXPRESSION DATA

0..* performer

ML identifier)

0..* expression components typeCode*: <=

expression levels)

pertinentInformation
typeCode*: <= PERT

0..* pertinentClinical

ClinicalPhenotype

Note:
Expression profile refers to both gene and protein expression levels.

Note:
The code attribute indicates in what molecule the variation occurs, i.e., DNA, RNA or Protein.

SequenceVariation

classCode*: <= OBS
moodCode*: <= EVN
id: II [0..1]
code: CD CWE [0..1] <= ActCode
text: FD IO 11